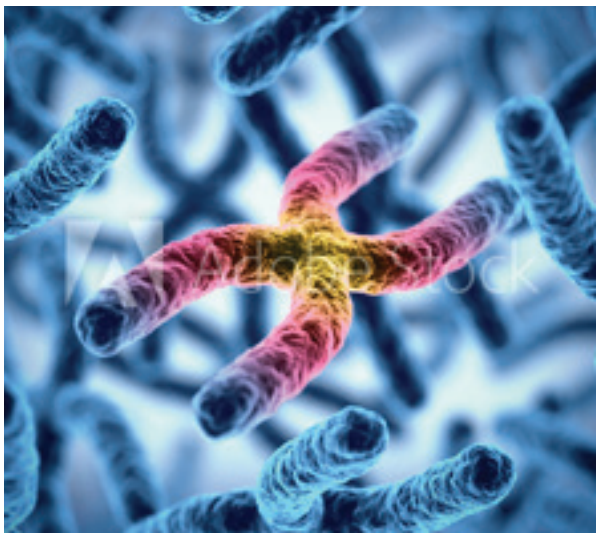
It's All About The Genes

By Richard Taavon, MD, FACOG

Newborn babies inherit certain family characteristics from their parents; hair color, eye color, height, and blood type are passed through generations through specific genes. Unfortunately, genes can pass on genetic disease, even if the parents do not have symptoms. Individuals often come to genetic counseling with an estimate of their risk of having a genetic disease and having a child affected with a genetic disorder. They may have been misdiagnosed by their practitioners, because misunderstanding of genetic principles is common and knowledge is advancing.

Most genes have two copies, one from their mother and the second from their father. A "carrier" is an individual who has a defect on one copy of a specific gene but not on the other. Therefore, by having one normal gene they do not have symptoms of a particular disease. When two "carriers" of a defective gene produce a child, there is a 25% chance that the child will inherit the disease by receiving an abnormal copy of the gene from each parent. There is also a 50% chance their child will be a carrier, and a 25% chance that their child will have a normal gene pair. Traditional carrier screening has generally targeted specific conditions with (1) fetal, neonatal, or early childhood onset, (2) a well-defined physical make-up, (3) physical or cognitive disabilities or a requirement for lifelong medical therapy. Prevalence, carrier frequency, detection rates, and residual risks are taken into account. Screening has often been limited to individuals at increased risk of these conditions because of ethnicity, race, or family history. Genetic screening tests can determine if an individual is a carrier for certain genetic disorders.

Various racial and ethnic groups are at higher risk to carry a specific abnormal gene. Sickle cell disease is more prevalent in African-American's, cystic fibrosis among Caucasian's, thalassemia among Greek, Italian, Mediterranean, or southern Asian descent, and Tay-Sachs disease among those of Eastern European Jewish and French Canadian descent. Some single gene disorders, can be treated if detected early. Cystic fibrosis requires lifelong treatment, where other genetic diseases, such as spinal muscular atrophy, have no treatment options. Because many people are of mixed ancestry: it is more common to offer genetic carrier screening to all individuals. Knowing whether or not you carry an abnormal gene, can make a difference in your future child's health.



Candidates for genetic counseling include individuals suspected of having a heritable disease, at risk of developing a heritable disease, and individuals concerned about having an affected child based upon family history, age, or ethnicity. Referral for genetic counseling is appropriate for women of advanced age, teratogen exposure during pregnancy, certain ethnic backgrounds, presence or birth defects, chromosomal abnormality, or intellectual disability in a parent, or the child of a family member.

The American College of Obstetrics and Gynecology recommends that cystic fibrosis carrier screening be offered to all women of reproductive age, as one of the most commonly inherited genetic disorders. If both parents are carriers for the same single gene disorder, testing is available to determine if an embryo has the genetic disease.

Patients with a family history of a specific disease should receive testing to determine if they are a carrier. Genetic counselors can help advise patients which tests are needed. Genetic counseling should include information on the likelihood of developing the disease, the impact of the disease, and any options for modifying the impact of disease. However, people with no family history should also consider screening for gene disorders.

A 2012 study analyzed the gene testing results of over 7000 patients. They found that women, with female factor infertility, had a higher rate of being a carrier for a single gene defect compared to healthy female donors. Men, with male factor infertility, also had a higher rate of being a carrier. They concluded that given the high rate of carriers among all groups, providers should offer carrier screening to patients and sperm/ egg donors prior to any reproductive treatment. This screening should also be considered in couples with a history of recurrent pregnancy loss.

Expanded carrier screening is a rapidly advancing field. Testing for multiple genetic disorders is done by blood testing or even a saliva sample. There are many companies who offer expanded screening panels. It is important that you and your physician discuss the benefits and risks of genetic testing including the possibility of false positive and false negative results. Genetic information may have unique risks based upon the meaning attributed to a disorder being genetic and the value of the information for family members.

Having a healthy child is the ultimate goal when building your family. Prenatal genetic screenings are valuable tools which offer parents important information about themselves and how the test results may impact their future family. WJ

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